

# GenomeConnect (The ClinGen Patient Registry) & Patient Registry Partnerships

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@GenomeConnect

@Savatt\_Juliann

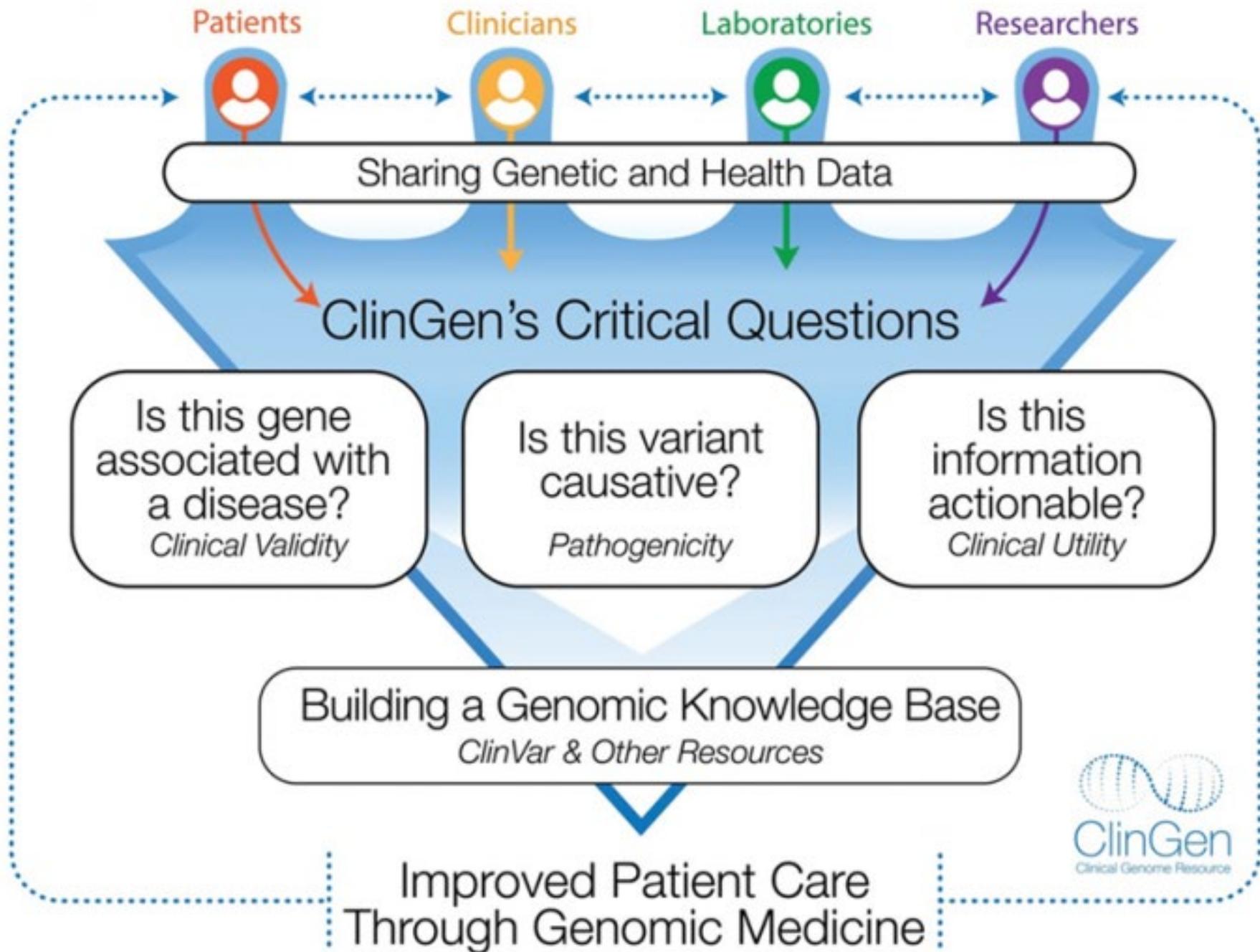


# Disclosures

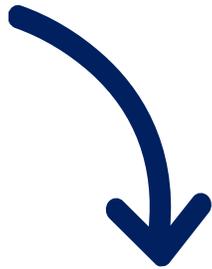
No conflicts of interest to disclose.

# Outline

- Explore how patients can share genetic and health information through patient registries.
- Describe how patient shared data can support and aid in variant classification and gene curation efforts.
- Describe how patient data sharing can provide patients with external opportunities for research, updates to their genetic results, and connections with other patients and families.



# GenomeConnect – ClinGen Patient Registry



Open to anyone who has had genetic testing

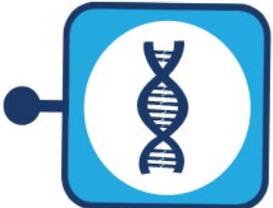
Sign-up and Consent Online



Upload genetic test report(s)



Provide health history via survey(s)\*

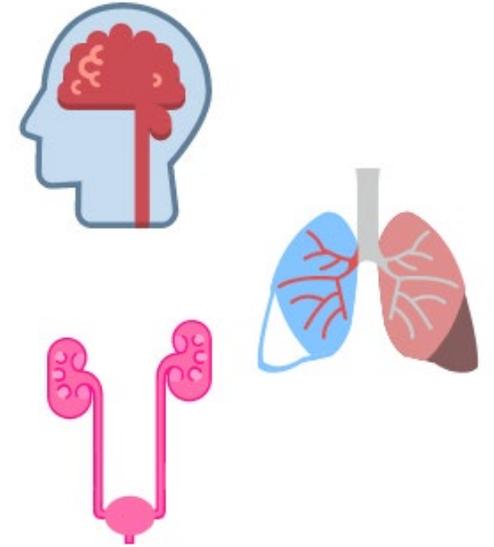


**GenomeConnect**  
The ClinGen Patient Portal

\* Additional surveys can be assigned based on the participants' initial responses

# GenomeConnect Health Survey

- Survey asks about general issues across 17 health systems.
- Each question provides examples in patient-friendly language.
- More specific questions are then assigned based on any systems or categories the participant had/has an issue with.



# GenomeConnect Health Survey

**Short stature** - Height is much shorter than is typical for your family and/or the average height for someone of your same age and sex



HP:0004322

human phenotype ontology

Tools ▾ Downloads ▾ Help ▾

No. Descendants Hierarchy ⓘ

- Abnormality of body height
  - Growth delay
  - Short stature
    - Disproportionate short stature
    - Proportionate short stature
    - Birth length less than 3rd percentile
    - Asymmetric short stature
    - Pituitary dwarfism

# GenomeConnect Collection of Genetic Data



**SCN8A**  
**c.2287A>G p.Ile763Val**  
**Likely Pathogenic**

**De novo**

**GeneDx**  
11/9/2015



Sequence Variant 1
Genome Browser Build
Transcript
Gene
Variant Classification
Variant name (c.) (ex:c.157T>G)
Variant name (p.) (ex:p.34Cys>Tyr)

# Data Sharing



Genetic and health data are de-identified and shared with NCBI's ClinVar



```
ACTGATGGTATGGGGCCAAGAGATATATCT  
CAGGTACGGCTGTCATCACTTAGACCTCAC  
CAGGGCTGGGCATAAAAGTCAGGGCAGAGC  
CCATGGTGCATCTGACTCCTGAGGAGAAGT  
GCAGGTTGGTATCAAGGTTACAAGACAGGT  
GGCACTGACTCTCTCGCCTATTGGTCTAT
```

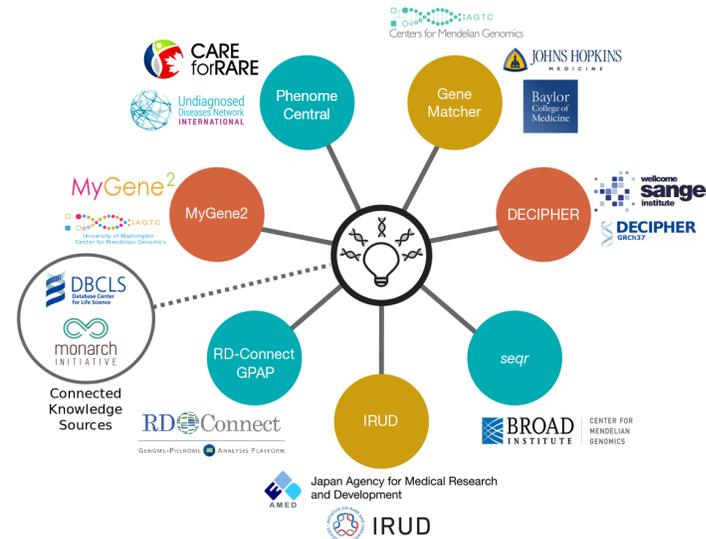
## ClinVar

ClinVar aggregates information about genomic variation and its relationship to human health.

# Data Sharing



Candidate Genes  
are submitted to  
Matchmaker  
Exchange

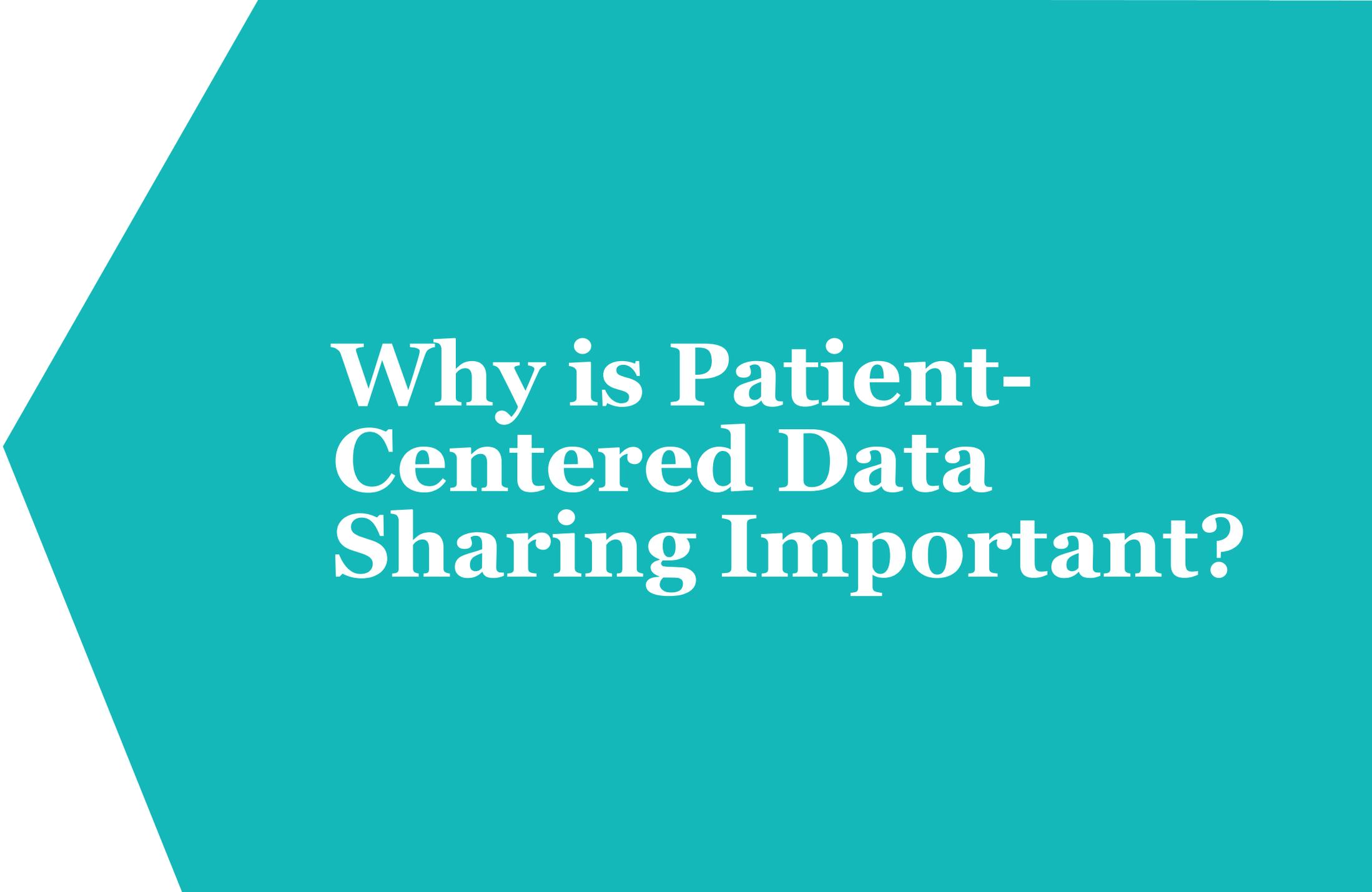


Candidate Gene

*Reclassification  
based on  
additional data*

No Association  
with Mendelian  
Disease

Association with  
Mendelian  
Disease

A large teal arrow pointing to the right, serving as a background for the text.

# **Why is Patient- Centered Data Sharing Important?**

# Benefits of Patient Data Sharing



Informing Variant Classification and Gene-Disease Validity



Facilitating Genetic Updates



Supporting Connections Between Stakeholders

# Benefits of Patient Data Sharing



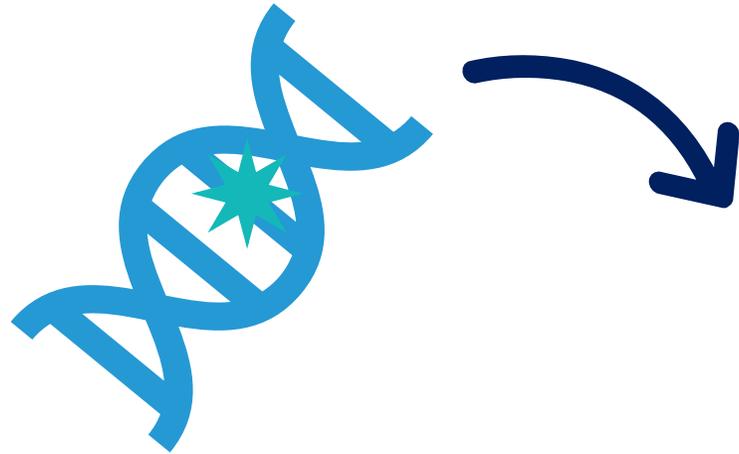
Informing Variant Classification and Gene-Disease Validity



Facilitating Genetic Updates



Supporting Connections Between Stakeholders



**ARTICLE**

**Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource**

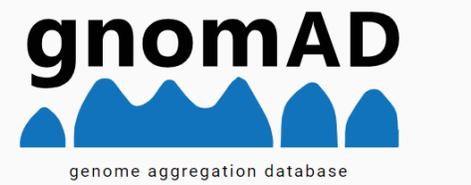
Natasha T. Strande,<sup>1,14</sup> Erin Rooney Riggs,<sup>2,14</sup> Adam H. Buchanan,<sup>3</sup> Ozge Ceyhan-Birsoy,<sup>4,5,6,7</sup> Marina DiStefano,<sup>4</sup> Selina S. Dwight,<sup>8</sup> Jenny Goldstein,<sup>1</sup> Rajarshi Ghosh,<sup>9</sup> Bryce A. Seifert,<sup>1</sup> Tam P. Sneddon,<sup>8</sup> Matt W. Wright,<sup>8</sup> Laura V. Milko,<sup>1</sup> J. Michael Cherry,<sup>8</sup> Monica A. Giovanni,<sup>3</sup> Michael F. Murray,<sup>3</sup> Julianne M. O'Daniel,<sup>1</sup> Erin M. Ramos,<sup>10</sup> Avni B. Santani,<sup>11,12</sup> Alan F. Scott,<sup>13</sup> Sharon E. Plon,<sup>9</sup> Heidi L. Rehm,<sup>4,5,6,7</sup> Christa L. Martin,<sup>2,3,\*</sup> and Jonathan S. Berg<sup>1,\*</sup>

With advances in genomic sequencing technology, the number of reported gene-disease relationships has rapidly expanded. However, the evidence supporting these claims varies widely, confounding accurate evaluation of genomic variation in a clinical setting. Despite the critical need to differentiate clinically valid relationships from less well-substantiated relationships, standard guidelines for such evaluation do not currently exist. The NIH-funded Clinical Genome Resource (ClinGen) has developed a framework to define and evaluate the clinical validity of gene-disease pairs across a variety of Mendelian disorders. In this manuscript we describe a proposed framework to evaluate relevant genetic and experimental evidence supporting or contradicting a gene-disease relationship and the subsequent validation of this framework using a set of representative gene-disease pairs. The framework provides a semi-quantitative measurement for the strength of evidence of a gene-disease relationship that correlates to a qualitative classification: "Definitive," "Strong," "Moderate," "Limited," "No Reported Evidence," or "Conflicting Evidence." Within the ClinGen structure, classifications derived with this framework are reviewed and confirmed or adjusted based on clinical expertise of appropriate disease experts. Detailed guidance for utilizing this framework and access to the curation interface is available on our website. This evidence-based, systematic method to assess the strength of gene-disease relationships will facilitate more knowledgeable utilization of genomic variants in clinical and research settings.

Is the gene associated with disease?



Is the variant pathogenic?



gnomAD v3 released! 71,702 genomes aligned on GRCh38.

**HHS Public Access**  
Author manuscript  
*Hum Mutat.* Author manuscript; available in PMC 2019 November 01.  
Published in final edited form as:  
*Hum Mutat.* 2018 November ; 39(11): 1581–1592. doi:10.1002/humu.23636.

**Gene-Specific Criteria for PTEN Variant Curation: Recommendations from the ClinGen PTEN Expert Panel**  
Jessica L. Mester<sup>1</sup>, Rajarshi Ghosh<sup>2</sup>, Tina Pesaran<sup>3</sup>, Robert Huether<sup>4</sup>, Rachid Karam<sup>5</sup>, Kathleen S. Hruska<sup>1</sup>, Helio A. Costa<sup>6</sup>, K. Sloan<sup>9,10</sup>, Kaitlin Sesock<sup>11</sup>, Felicia Hernandez<sup>12</sup>, Madhuri Hegde<sup>14,15</sup>, and Charis Teruya-Feldstein<sup>13</sup>

**Technical standards for the interpretation of constitutional copy-number variants: a recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen)**  
Erin Rooney Riggs, MS, CGC<sup>1</sup>, Erica F. Andersen, PhD<sup>2,3</sup>, Athena M. Cherry, PhD<sup>4</sup>, Sibel Kantarci, PhD<sup>5</sup>, Hutton Kearney, PhD<sup>6</sup>, Ankita Patel, PhD<sup>7</sup>, Gordana Raca, MD, PhD<sup>8</sup>, Deborah I. Ritter, PhD<sup>9</sup>, Sarah T. South, PhD<sup>10</sup>, Erik C. Thorland, PhD<sup>5</sup>, Daniel Pineda-Alvarez, MD<sup>11</sup>, Swaroop Aradhya, PhD<sup>12</sup> and Christa Lese Martin, PhD<sup>13</sup>

**GUIDELINE**      Open Access

**Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework**

Sarah E. Brnich<sup>1</sup>, Ahmad N. Abou Tayoun<sup>2</sup>, Fergus J. Couch<sup>3</sup>, Garry R. Cutting<sup>4</sup>, Marc S. Greenblatt<sup>5</sup>, Christopher D. Heinen<sup>6</sup>, Dona M. Kanavy<sup>1</sup>, Xi Luo<sup>7</sup>, Shannon M. McNulty<sup>1</sup>, Lea M. Starita<sup>8,9</sup>, Sean V. Tavtigian<sup>10</sup>, Matt W. Wright<sup>11</sup>, Steven M. Harrison<sup>12</sup>, Leslie G. Biesecker<sup>13</sup>, Jonathan S. Berg<sup>1\*</sup> and On behalf of the Clinical Genome Resource Sequence Variant Interpretation Working Group

# Patient Data Sharing Can Inform Variant Classification and Gene-Disease Validity

- Can provide access to unpublished cases to inform gene curation.
- Offers case-level information to inform variant classification.



## Gene-Disease Validity

Can variation in this gene cause disease?

[Learn More](#)

[Browse Curations](#) 



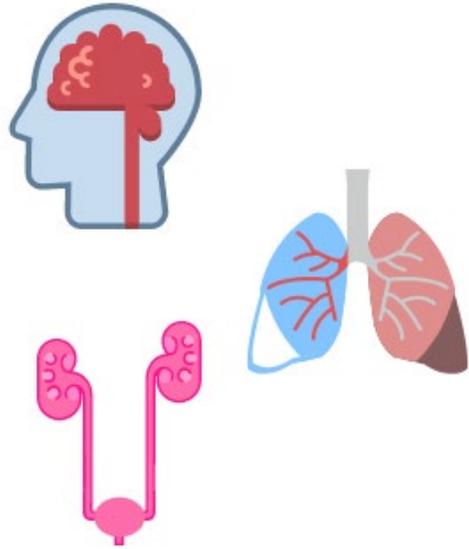
## Variant Pathogenicity

Which changes in the gene cause disease?

[Learn More](#)

[Browse Curations](#) 

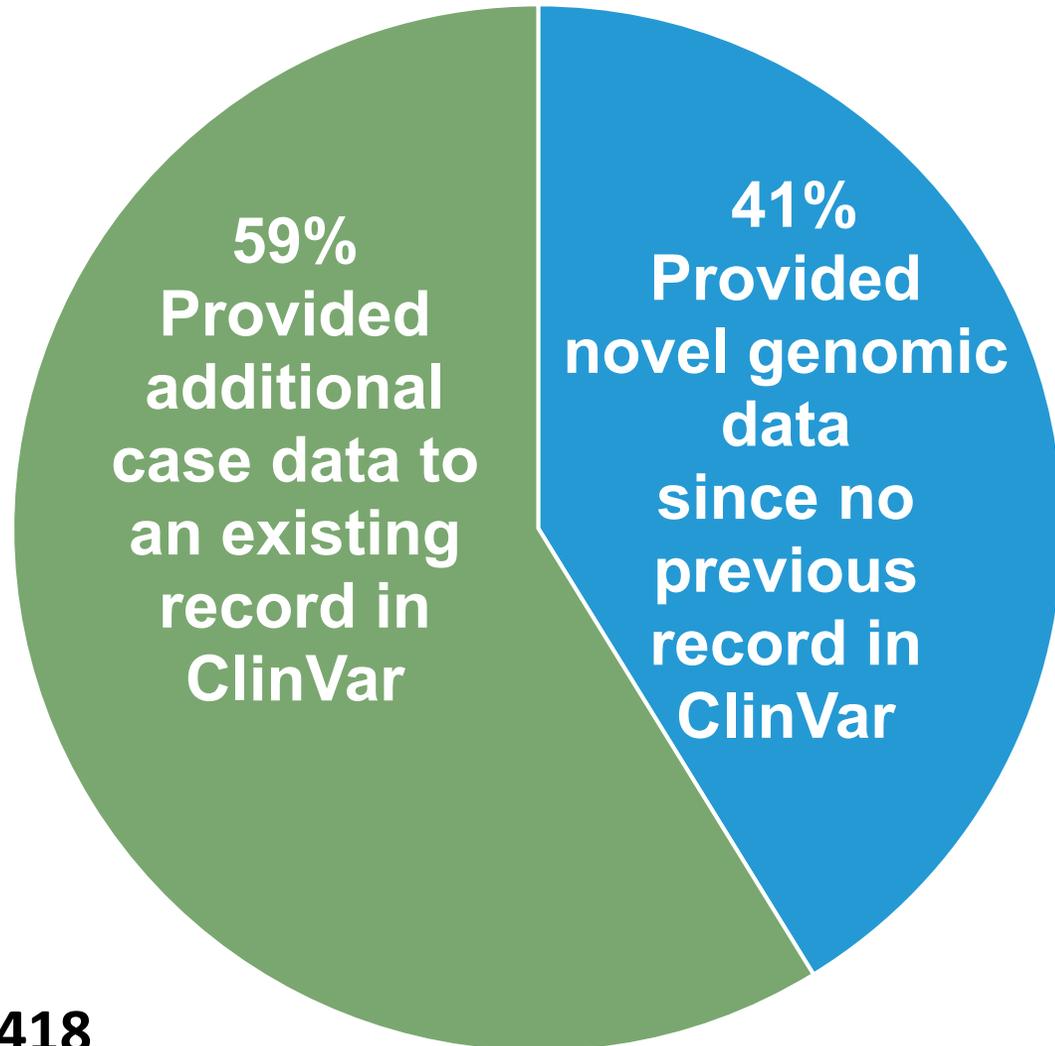
# Contributing Additional Phenotypic Data



Patients provide enhanced phenotypic data, information regarding inheritance of the variant, and the ability to re-contact for additional information, as needed.

# Sharing Novel Genomic Data

## ClinVar Submissions to Date



n=1418

Patients provide genomic variants that are not otherwise available which can inform our understanding of genetic changes and of gene-disease relationships.

# Case Example 1



GenomeConnect  
The ClinGen Patient Portal

**SCN8A**  
**c.2287A>G p.Ile763Val**  
**Likely Pathogenic**

**De novo**

**LAB1**  
11/9/2015

*SCN8A* is associated with infantile epileptic encephalopathy

# Case Example 1

## ClinVar



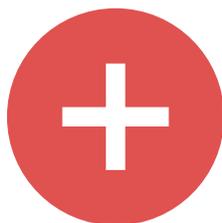
### LAB 1

### LAB 2

**SCN8A**  
c.2287A>G p.Ile763Val  
**Likely Pathogenic**

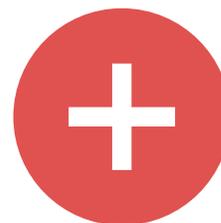
*de novo*

**LAB1**  
11/9/2015



**SCN8A**  
c.2287A>G p.Ile763Val  
**Likely Pathogenic**

**LAB1**  
11/9/2015



**SCN8A**  
c.2287A>G p.Ile763Val  
**Uncertain**

**LAB2**  
6/3/2014



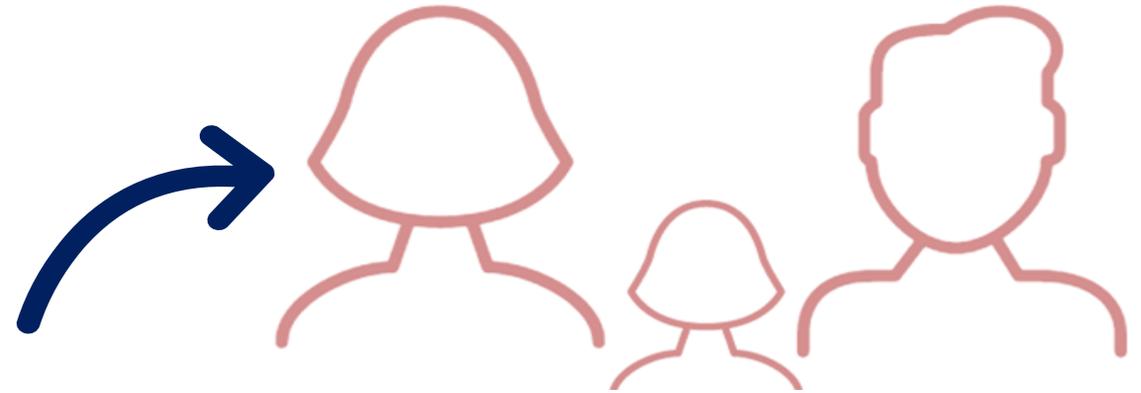
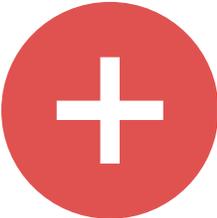
# Case Example 1

**LAB 2**

*Update*

**SCN8A**  
**c.2287A>G p.Ile763Val**  
**Likely Pathogenic**

**Lab2**  
**3/2/2018**



# Benefits of Patient Data Sharing



Informing Variant Classification and Gene-Disease Validity

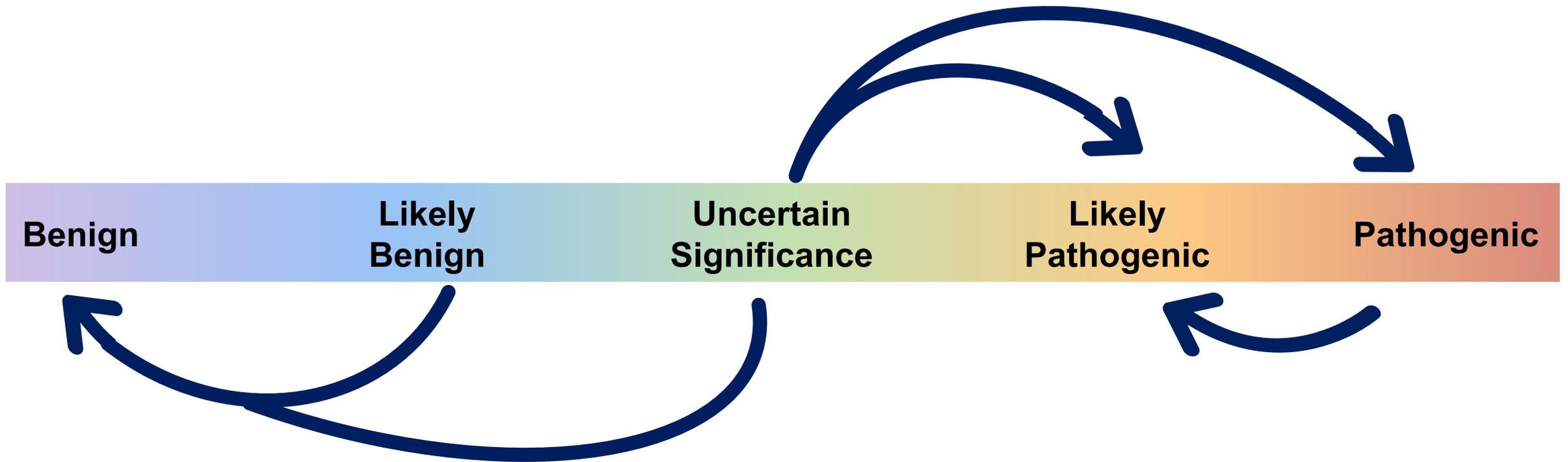


Facilitating Genetic Updates



Supporting Connections Between Stakeholders

# Variant Reclassifications



At just one clinical laboratory, between 2006 and 2016, 6.4% variants were reclassified leading to 59,955 amended reports.

(Mersch et al., 2018)

# Variant Reclassifications - Recontact

European Journal of Human Genetics (2019) 27:169–182  
https://doi.org/10.1038/s41431-018-0285-1

ESHG

POLICY



## Recontacting patients in clinical genetics services: recommendations of the European Society of Human Genetics

Daniele Carrieri<sup>1</sup> · Heidi C. Howard<sup>2</sup> · Caroline Benjamin<sup>3,4</sup> · Angus J. Clarke<sup>5</sup> · Sandi Dheensa<sup>6</sup> · Shane Doheny<sup>5</sup> · Naomi Hawkins<sup>7</sup> · Tanya F. Halbersma-Konings<sup>8</sup> · Leigh Jackson<sup>9</sup> · Hülya Kayserili<sup>10</sup> · Susan E. Kelly<sup>1</sup> · Anneke M. Lucassen<sup>6,11</sup> · Álvaro Mendes<sup>12</sup> · Emmanuelle Rial-Sebbag<sup>13</sup> · Vigdís Stefánsdóttir<sup>14</sup> · Peter D. Turnpenny<sup>15</sup> · Carla G. van El<sup>16</sup> · Irene M. van Langen<sup>8</sup> · Martina C. Cornel<sup>16</sup> · Francesca Forzano<sup>17</sup> ·  
On behalf of the European Society of Human Genetics

### 7. Recontacting should be a shared responsibility with patients

Where patients relocate or change their mind in respect of being recontacted, it is important for the genetics service to be kept informed. Placing additional responsibility on patients to initiate recontacting might be seen as a pragmatic solution to the current lack of resources and infrastructure but risks distracting services from their most important tasks and reinforcing inequities in health care. Any patient-led recontacting requires an effective partnership between patient and genetic services, in which the patient is offered education on why, how and when s(he) might consider establishing a new contact with a genetics centre.

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ACMG POLICY STATEMENT

Genetics  
inMedicine



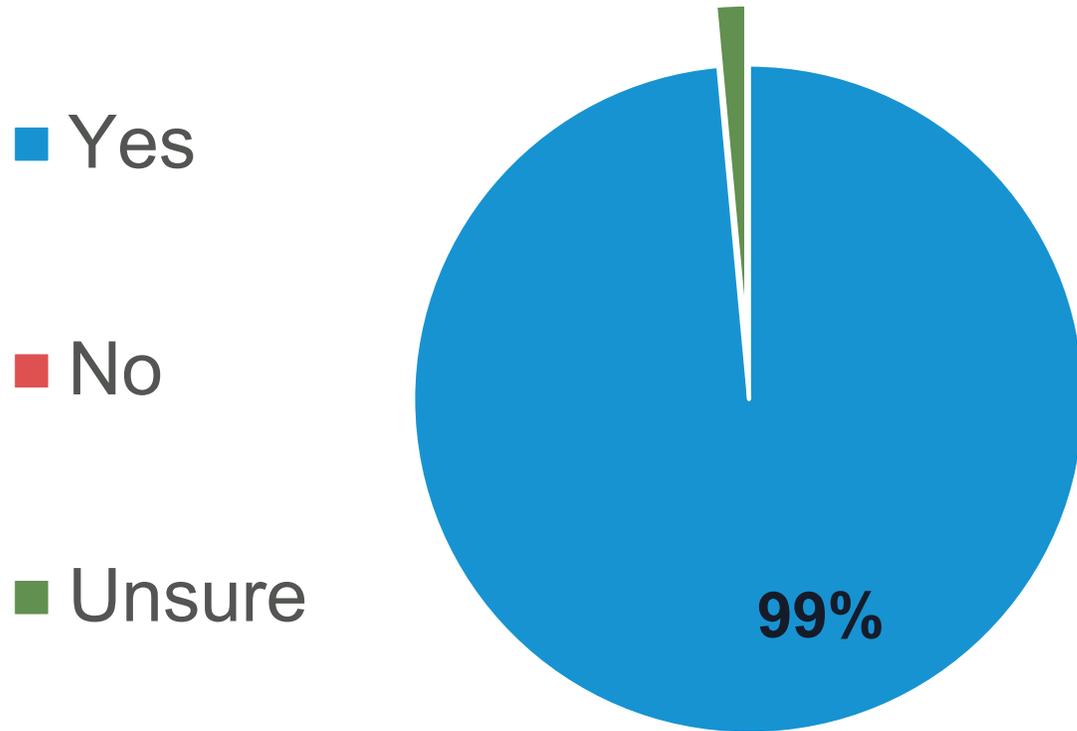
## Patient re-contact after revision of genomic test results: points to consider—a statement of the American College of Medical Genetics and Genomics (ACMG)

3. The ordering provider should emphasize, through discussion and in written explanation to the patient, that the ordering provider cannot promise that re-contact regarding a revised interpretation will occur unless the patient initiates the re-contact.

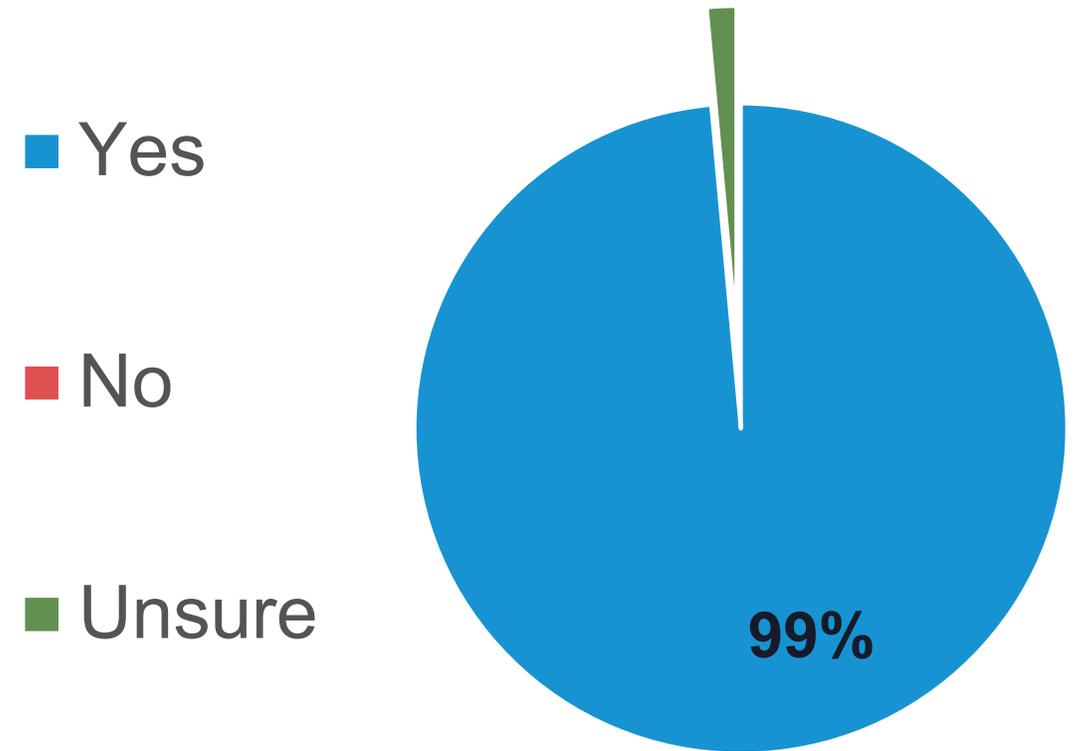
Patients share in the responsibility to remain up to date about their genetic test results.

(Carrieri et al., 2019; David et al., 2019)

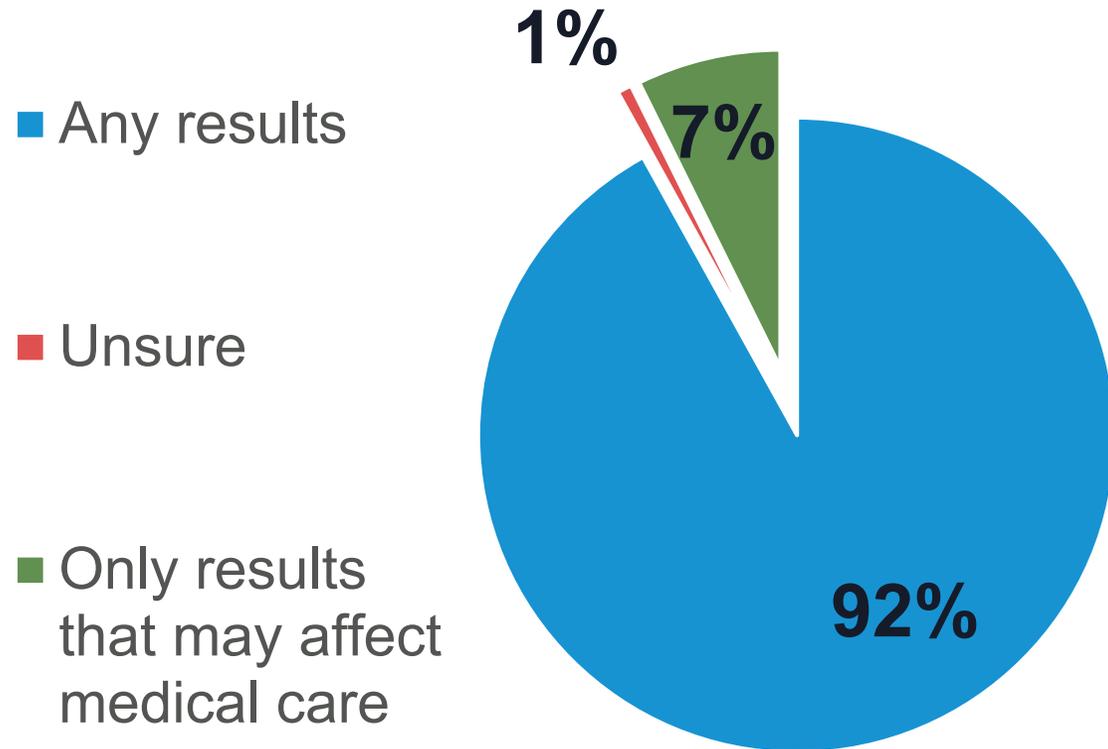
If your/ your family member's genetic testing results were updated, **would you want to learn about these updates?**



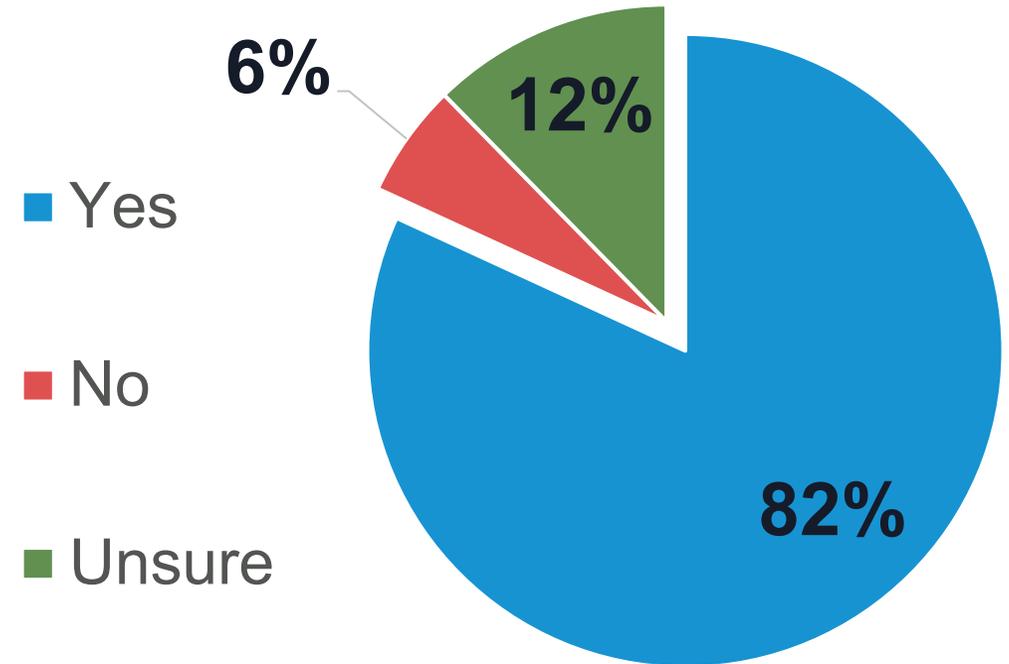
If **GenomeConnect** learned about these updates, would you want us to contact you with this information?



What **type of updates** would you like to receive if you were able to choose?

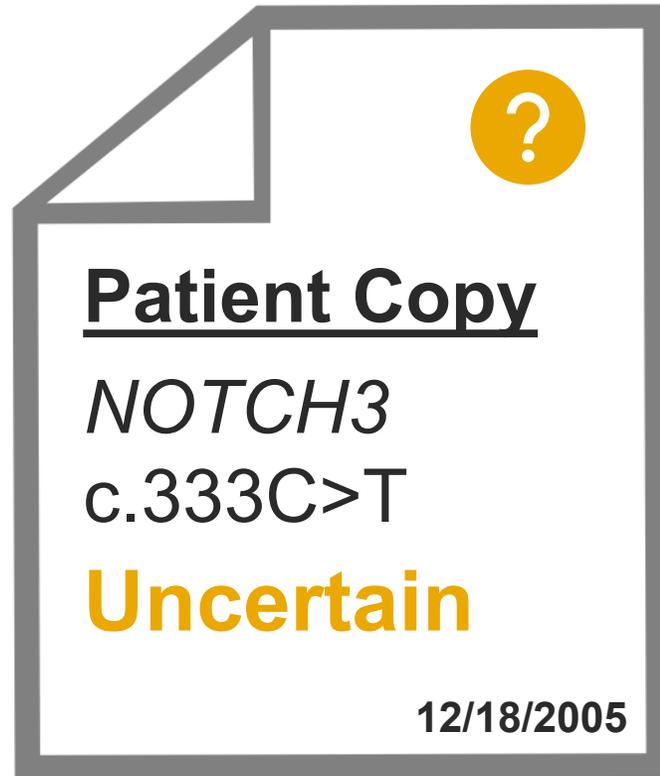


Regardless of your own views, do you think participants should have the **option to decline** receiving updates?

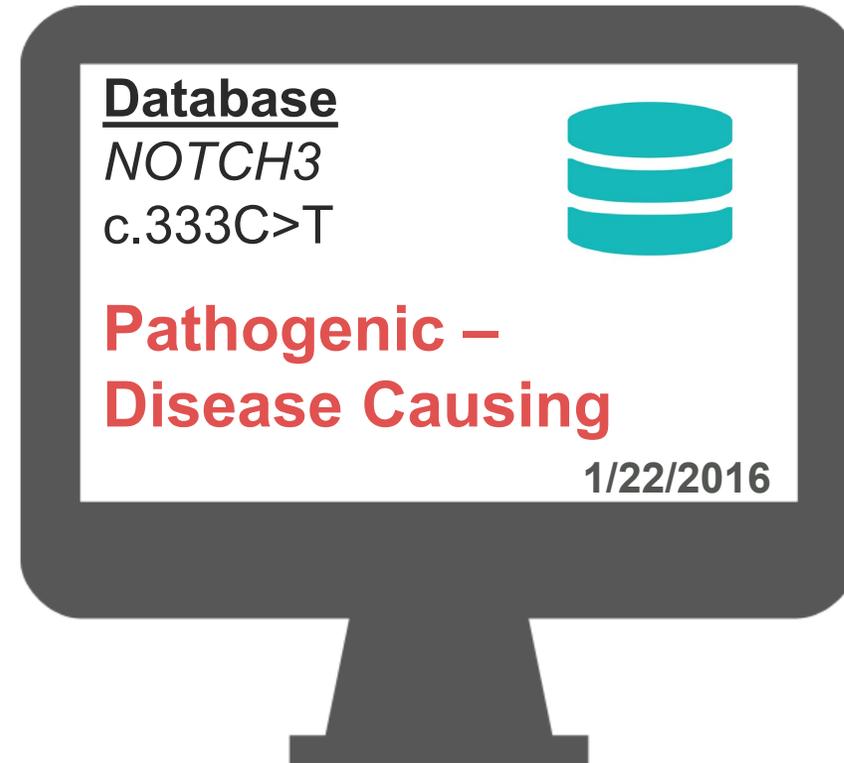


# Sharing Information Back with Participants

Participant  
Report

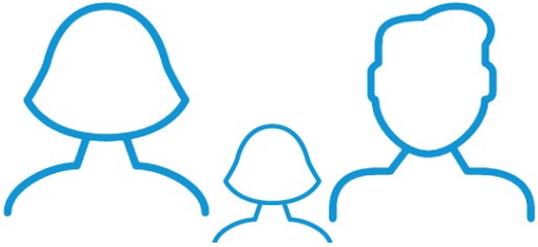


ClinVar Entry from  
Reporting Lab



Email to the participant informing them there might be an update to their results.

# Case Example 2

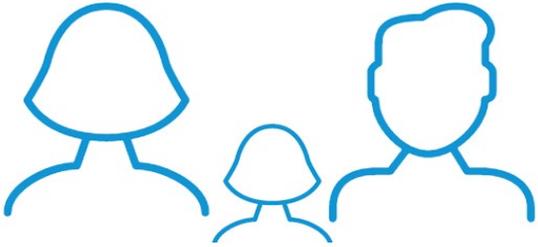


- **August 2016 - Patient underwent exome in due to a history of a seizures and intellectual disability.**
- **Family enrolled in GenomeConnect and gene was submitted to Matchmaker Exchange.**



*CSNK2B*  
*de novo*  
**Uncertain**

# Case Example 2



**December 2019 – GenomeConnect became aware of a relationship between *CSNK2B* and mild to profound intellectual disabilities, developmental delays, and various types of seizures.**

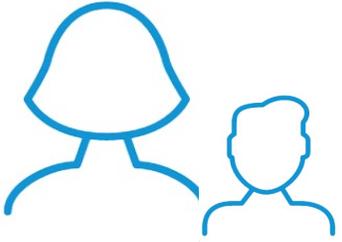
- Contacted the reporting laboratory
  - Upgraded the *CSNK2B* gene to disease-associated, and recently reported the same variant out as pathogenic



*CSNK2B*  
c.367+5delG  
*de novo*  
**Pathogenic**

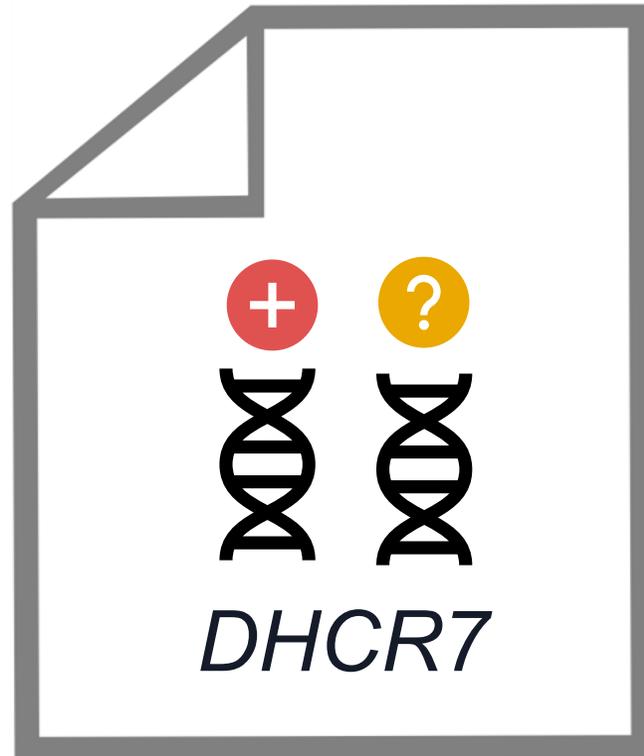
**December 2019 – Family was contacted by GenomeConnect and received an updated report from the laboratory.**

# Case Example 3

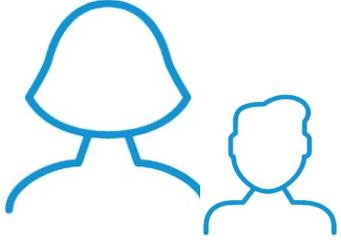


**Participant underwent exome sequencing in 2015 due to a history of prenatal cystic hygroma, behavioral issues, fine motor delay, and dysmorphic features.**

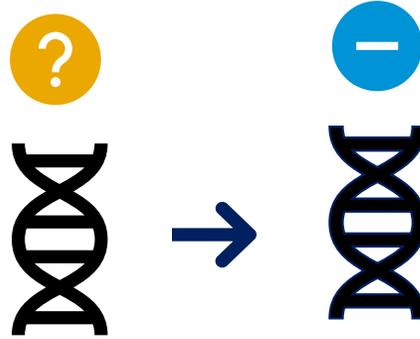
- No causative variant but two variants in *DHCR7* identified.



# Case Example 3



**GenomeConnect submitted to ClinVar and learned that the VUS had been reclassified to Likely Benign by the reporting laboratory.**



*DHCR7*

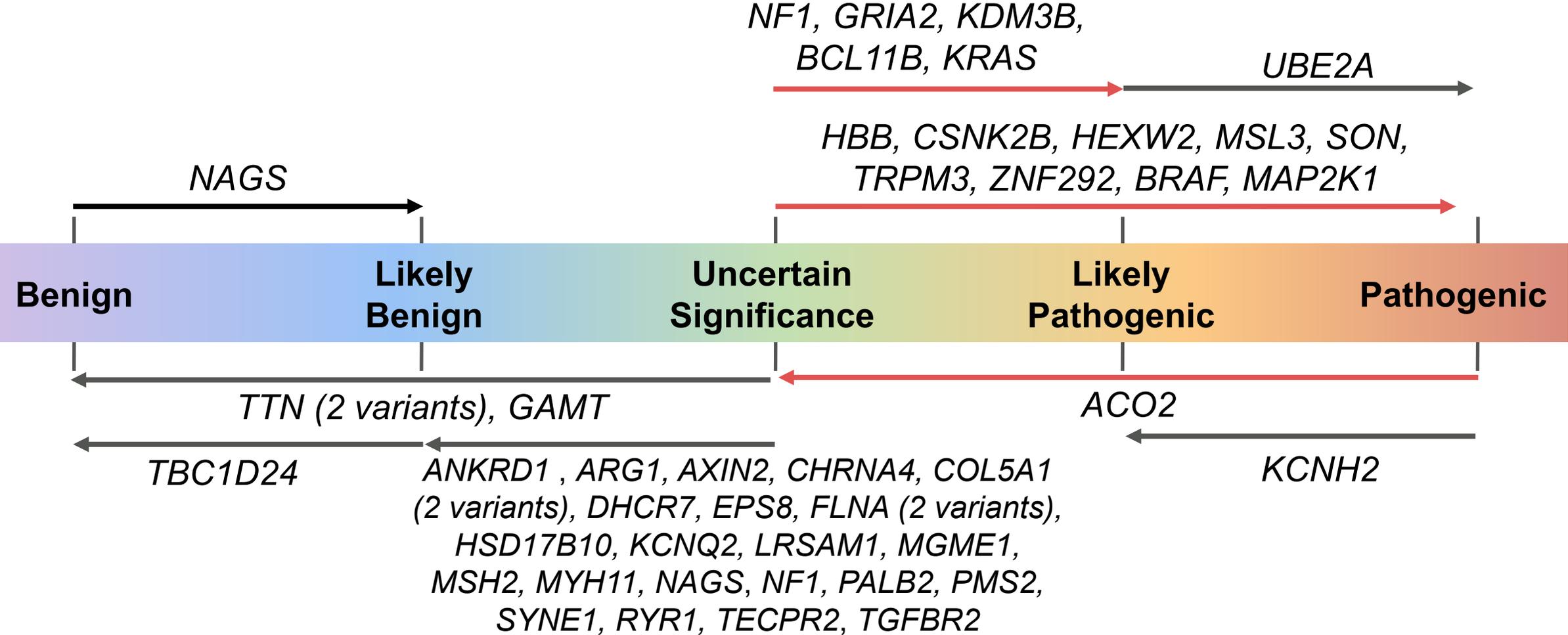
**Recontacted the family who then underwent exome reanalysis and received an updated report.**

- Documented reclassification
- Identified a novel, *de novo* VUS in a gene associated with dysmorphic features, neurodevelopmental disorders, and congenital anomalies.

# Updates Identified & Shared

**47 Updates Identified and Shared**

**15 Updates Likely to Impact Diagnosis and/or Care**



# Benefits of Data Sharing



Informing Variant Classification and Gene-Disease Validity



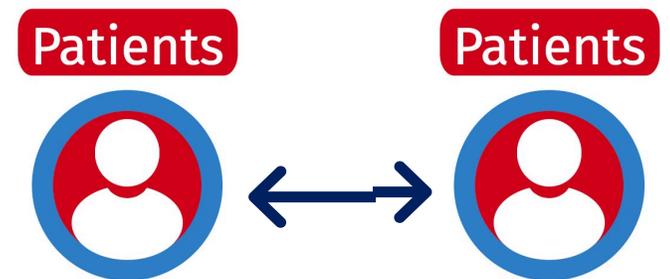
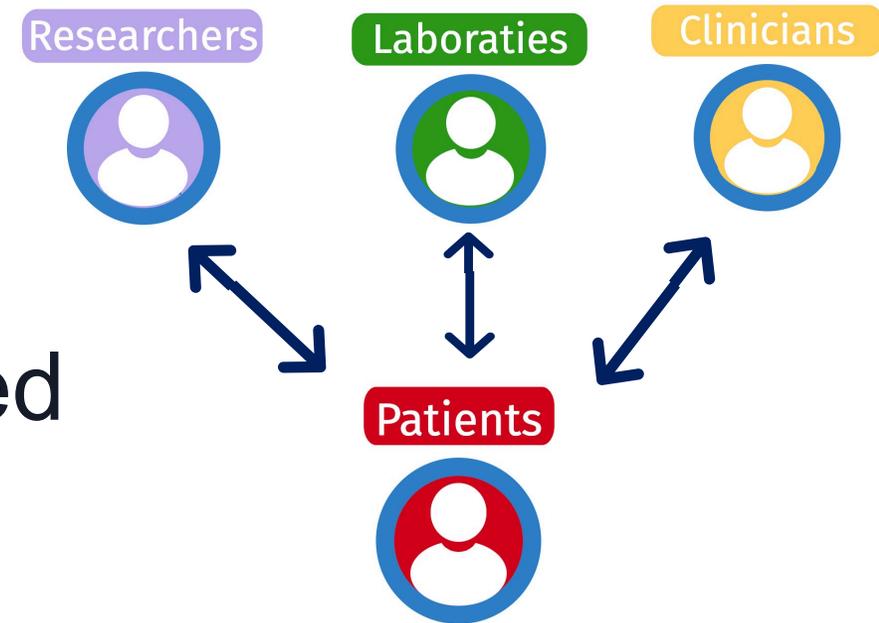
Facilitating Genetic Updates



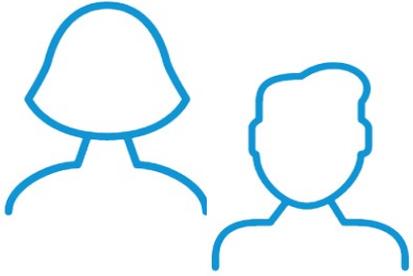
Supporting Connections Between Stakeholders

# Enabling Connections with Other Stakeholders

- Enable connections with researchers or clinicians interested in a particular gene or variant
- Participants can match with one another based on gene, disease, or geographic location



# Case Example 4

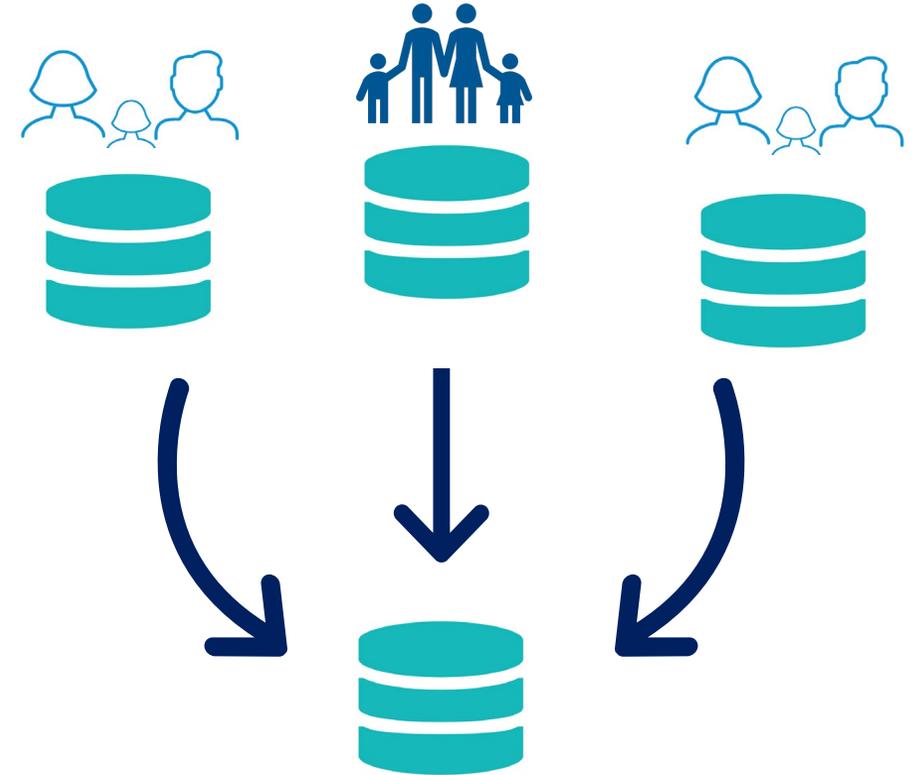


- **March 2016 - Patient underwent ES due to history of ASD, ID, Tourette syndrome, hypotonia.**
  - Two candidate genes were reported including *GRIA1*.
- **2019 – Patient enrolled in GenomeConnect and GenomeConnect submitted to Matchmaker Exchange**
- **June 2020**
  - Researcher collecting case series with variants in *GRIA1*
  - Connected family with researcher for inclusion in publication

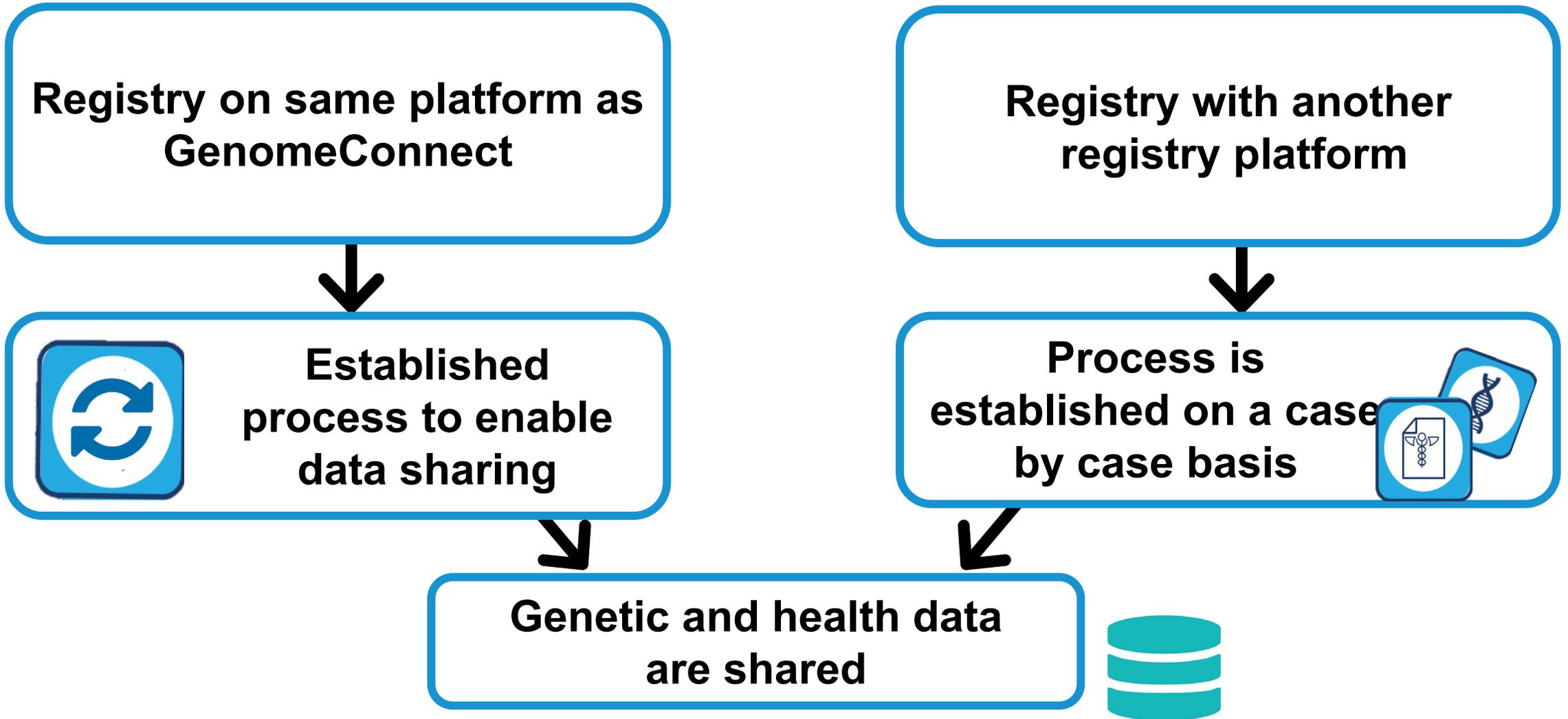
# ClinGen Patient Data Sharing Program

# GenomeConnect – Patient Data Sharing Program

- Other registries are and will be collecting valuable genetic and health data that is may not be publicly shared.
- GenomeConnect team created the Patient Data Sharing Program to afford more patients the opportunity to share data



# GenomeConnect – Patient Data Sharing Program



# Patient Data Sharing Program

SIMONS  
SEARCHLIGHT



**CFC International**  
Cardio-Facio-Cutaneous Syndrome

**NO STOMACH  
FOR CANCER**<sup>®</sup>  
Supporting Research. Empowering Families.

**GM1  
PATIENT NETWORK**

ASSOCIATION FOR  
**CREATINE**  
DEFICIENCIES

EDUCATION ADVOCACY RESEARCH

  
**Costello  
Syndrome**  
**FAMILY NETWORK**



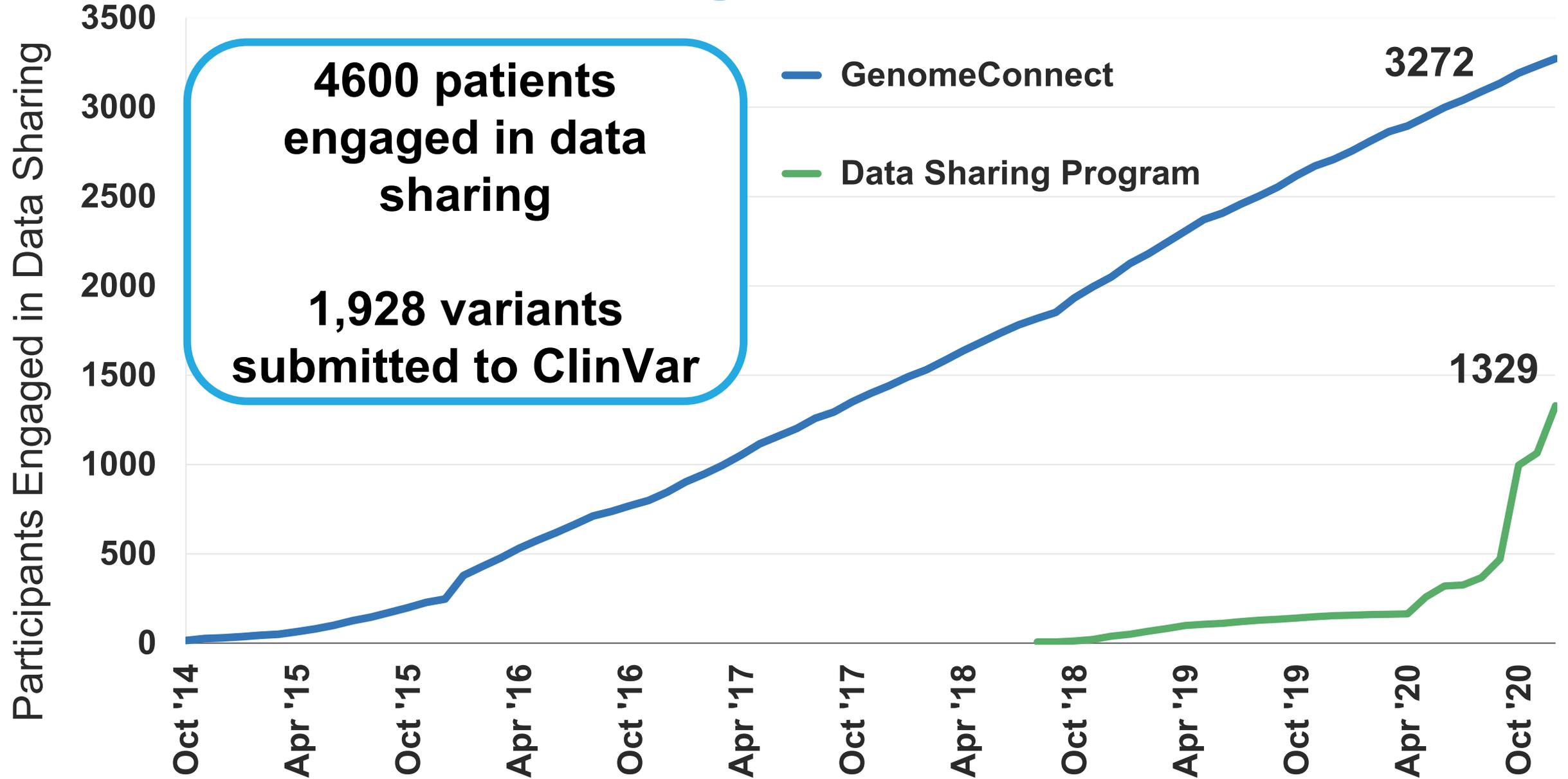
INVITAE



Grace Science Foundation  
NGLY1 deficiency syndrome

**HNDS**

# Patient Data Sharing



# Patient Data Sharing

**ClinGen is supporting data sharing from around the world.**

**10% of participants are from outside the US.**

**Participants enrolled in data sharing are from 52 countries.**



# How to Get Involved

# How to Get Involved

- Use patient shared data to inform understanding of the relationship between genes, variants, and health.
- Variant information from GenomeConnect participants or other registries engaging in data sharing is submitted to ClinVar periodically.
  - Information regarding the variant and phenotype are included in the submission.
  - Contact us with additional questions.



## Variant Pathogenicity

Which changes in the gene cause disease?

[Learn More](#) [Browse Curations](#)



## Gene-Disease Validity

Can variation in this gene cause disease?

[Learn More](#) [Browse Curations](#)



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[Learn More](#) [Browse Curations](#)



## Gene-Disease Validity

Can variation in this gene cause disease?

[Learn More](#) [Browse Curations](#)



# NM\_015047.3(EMC1):c.245C>T (p.Thr82Met)

[Cite this record](#)

**Interpretation:** Uncertain significance

**Review status:** ★★☆☆ criteria provided, multiple submitters, no conflicts

**Submissions:** 4 (Most recent: May 3, 2020)

**Last evaluated:** May 3, 2020

**Accession:** VCV000219100.3

**Variation ID:** 219100

**Description:** single nucleotide variant

## Variant details

[Conditions](#)[Gene\(s\)](#)

FEEDBACK

### NM\_015047.3(EMC1):c.245C>T (p.Thr82Met)

**Allele ID:** 216792

**Variant type:** single nucleotide variant

**Variant length:** 1 bp

**Cytogenetic location:** 1p36.13

**Genomic location:** 1: 19243991 (GRCh38) [GRCh38](#) [UCSC](#)  
1: 19570485 (GRCh37) [GRCh37](#) [UCSC](#)

#### HGVS:

Nucleotide	Protein	Molecular consequence
<a href="#">NC_000001.10:g.19570485G&gt;A</a>		
<a href="#">NC_000001.11:g.19243991G&gt;A</a>		
<a href="#">NM_001271427.2:c.245C&gt;T</a>	NP_001258356.1:p.Thr82Met	missense

[... more HGVS](#)

**Protein change:** T82M

**Other names:** -

**Canonical SPDI:** [NC\\_000001.11:19243990:G:A](#)

**Functional consequence:** -

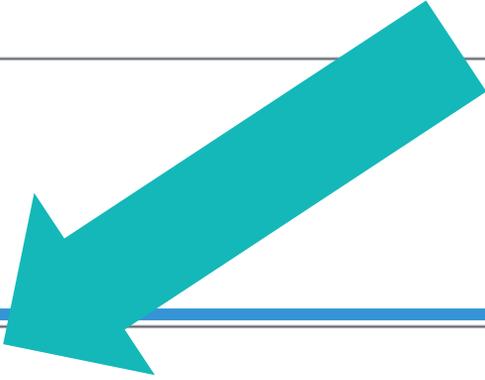
**Global minor allele frequency (GMAF):** -

**Allele frequency:** The Genome Aggregation Database (gnomAD), exomes 0.00002

**Links:** [dbSNP: rs869320625](#)  
[ClinGen: CA358733](#)  
[UniProtKB: Q8N766#VAR\\_076915](#)  
[OMIM: 616846.0003](#)

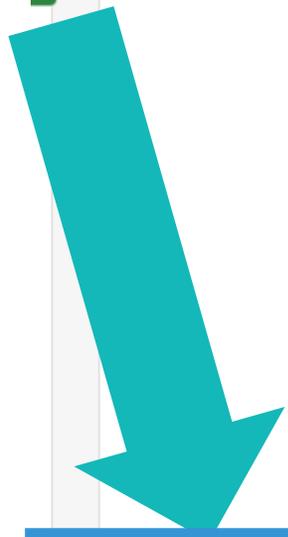
Interpretation (Last evaluated)	Review status (Assertion criteria)	Condition (Inheritance)	Submitter	Supporting information (See all)
Uncertain significance (-)	criteria provided, single submitter (Harel et al. (Am J Hum Genet. 2016)) Method: research	not specified (Autosomal recessive inheritance) Allele origin: germline	Lupski Lab, Baylor-Hopkins CMG, Baylor College of Medicine Accession: SCV000258466.1 Submitted: (Jan 11, 2016)	Evidence details Publications PubMed (1)
Uncertain significance (May 03, 2020)	criteria provided, single submitter (ACMG Guidelines, 2015) Method: clinical testing	Cerebellar atrophy, visual impairment, and psychomotor retardation Allele origin: unknown	Genomic Research Center, Shahid Beheshti University of Medical Sciences Accession: SCV000746362.2 Submitted: (May 03, 2020)	Evidence details
Pathogenic (Mar 29, 2016)	no assertion criteria provided Method: literature only	CEREBELLAR ATROPHY, VISUAL IMPAIRMENT, AND PSYCHOMOTOR RETARDATION Allele origin: germline	OMIM Accession: SCV000266489.1 Submitted: (Mar 29, 2016)	Evidence details Publications PubMed (1)
not provided (-)	no assertion provided Method: phenotyping only	EMC1-Related Disorder Allele origin: paternal	GenomeConnect, ClinGen Accession: SCV000986832.1 Submitted: (Jan 30, 2019)	Evidence details Comment: Variant interpreted as Likely pathogenic and reported on 06/18/2018 by GTR ID 26957. GenomeConnect assertions are reported exactly as they appear on the patient-provided report from the testing laboratory. GenomeConnect staff make no attempt to reinterpret the clinical significance of the variant. (less)

FEEDBACK



#	SCV	Submitter	Allele origin	Clinical features (Affected status)	Age	Sex	Testing laboratory	Testing laboratory interpretation	Date interpretation reported to submitter
1	SCV000986832.1	GenomeConnect, ClinGen	paternal	Abnormality of the skull (yes) Oral-pharyngeal dysphagia (yes) Abnormality of eye movement (yes) Abnormality of vision (yes) Abnormality of the optic nerve (yes) Hyperacusis (yes) Sensorineural hearing loss (yes) Cerebral palsy (yes) Cognitive impairment (yes) Abnormality of coordination (yes) EEG abnormality (yes) Encephalopathy (yes) Hypertonia (yes) Generalized hypotonia (yes) Memory impairment (yes) Abnormality of movement (yes) Seizures (yes) Autistic behavior (yes) Pectus excavatum (yes) Hip dysplasia (yes) Abnormality of the curvature of the vertebral column (yes) Joint hypermobility (yes) Abnormality of facial musculature (yes) Abnormality of muscle physiology (yes) Abnormality of the musculature of the pelvis (yes) Arrhythmia (yes) Cardiomyopathy (yes) Abnormal pattern of respiration (yes) Abnormality of the upper respiratory tract (yes) Feeding difficulties (yes) Abnormality of esophagus morphology (yes) Abnormal number of teeth (yes)	0-9 years	female	GeneDx	Likely pathogenic	2018-06-18

FEEDBACK



 Choose Columns

Email our team with questions:  
[info@genomeconnect.org](mailto:info@genomeconnect.org)



**COVID-19 is an emerging, rapidly evolving situation.**



[Public health information \(CDC\)](#) | [Research information \(NIH\)](#) | [SARS-CoV-2 data \(NCBI\)](#) | [Prevention and treatment information \(HHS\)](#)

**Submitters and their submissions**

This page lists all submitters and the summary of their contributions, ordered inversely by the number of submissions. We acknowledge their support.

Note that the number of submissions for any submitter may be different from the number of variants found in a search based on the name of that submitter. A submission is for the interpretation of a variant and a condition; if the submitter has interpreted the variant with respect to more than one condition, then the count of submissions will be higher than the search result.

Submitter	Maximum review status	Total submissions	Submissions with interpretations	Total Genes	Last updated
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	392984	392981	12740	Oct 09, 2020
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	208455	208455	2818	Dec 18, 2020
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	121912	121778	26759	Nov 20, 2020
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	72593	72593	1714	Nov 30, 2020
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	45028	45028	2408	Sep 19, 2018
<a href="#">Genetic Services Laboratory; Illumina</a>	Assertion criteria	43495	43495	84	May 19, 2020
<a href="#">Hopkins University</a>	-	32029	32029	5266	Jan 21, 2021
<a href="#">Molecular Medicine; Partners HealthCare Personalized</a>	Assertion criteria	24235	24161	1751	Jun 03, 2020
<a href="#">Molecular Medicine; Partners HealthCare Personalized</a>	Assertion criteria	20976	20976	346	Aug 05, 2019
<a href="#">Molecular Medicine; Partners HealthCare Personalized</a>	Assertion criteria	20219	20219	6296	Aug 26, 2020
<a href="#">Integrated Genetics/Laboratory Corporation of America; Laboratory Corporation of America</a>	Assertion criteria	18507	18483	849	Dec 31, 2020
<a href="#">PreventionGenetics</a>	Assertion criteria	18417	18417	1562	Nov 12, 2020
<a href="#">Cincinnati Children's Hospital Medical Center Laboratory of Genetics and Genomics; Cincinnati Children's Hospital Medical Center</a>	Assertion criteria	17788	17788	19587	Apr 27, 2020
<a href="#">Natera, Inc.</a>	-	15814	15814	358	Dec 28, 2020
<a href="#">Genetic Services Laboratory; University of Chicago</a>	Assertion criteria	15337	15337	1256	Nov 01, 2020
<a href="#">Quest Diagnostics Nichols Institute San Juan Capistrano</a>	Assertion criteria	13711	13711	16434	Dec 31, 2020
<a href="#">Athena Diagnostics Inc</a>	Assertion criteria	13004	13004	806	Dec 30, 2020
<a href="#">ARUP Laboratories, Molecular Genetics and Genomics; ARUP Laboratories</a>	Assertion criteria	10404	10404	1005	Dec 11, 2020
<a href="#">Mendelics</a>	Assertion criteria	9265	9265	1772	Nov 23, 2020
<a href="#">Evidence-based Network for the Interpretation of Germline Mutant Alleles (ENIGMA); c/o QIMR Berghofer Medical Research Institute</a>	Expert panel	7456	7455	9	Aug 19, 2019
<a href="#">ISCA site 1</a>	-	7274	7274	31963	Feb 04, 2019
<a href="#">Database of Curated Mutations; Washington University in St Louis</a>	-	6947	6878	132	Jul 18, 2016
<a href="#">GeneReviews</a>	-	6260	6259	1642	Jan 15, 2021
<a href="#">Blueprint Genetics</a>	Assertion criteria	5274	5274	705	Nov 16, 2020

<http://bit.ly/ClinVarList>

# How to Get Involved

- Work with ClinGen to expand data sharing.
- If you work with patients, inform them of opportunities to share data through GenomeConnect.
- If you work with a patient registry, discuss the opportunity to engage in the Patient Data Sharing Program.



# Summary

- GenomeConnect and other patient registries offers a structured way for patients to share genotype and phenotype data.
- Patients are an important source of novel genomic data and phenotypic details to inform understanding of genetics.
- Patients engaged in data sharing can remain informed about their results, learn about additional research studies, and connect with other patients and families.
- ClinGen is working with other patient groups and registries to facilitate patient data sharing.

# Acknowledgements

## GenomeConnect at Geisinger

- Christa Lese Martin, PhD, FACMG
- Erin Rooney Riggs, MS, CGC
- Taylor Bingaman, BS
- Molly E Good, BA
- Lianna D Paul, BS
- Alexis Heidlebaugh, MS

## GenomeConnect at the Broad

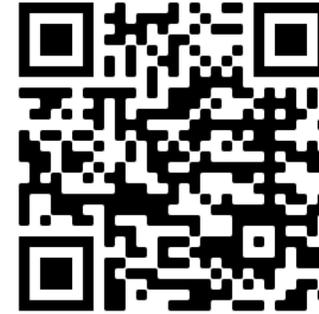
- Heidi Rehm, PhD
- Danielle Azzariti, MS, CGC

## Other Contributors

- David H. Ledbetter, PhD, FACMG
- W. Andy Faucett, MS, LGC
- Steven Harrison, PhD (ClinGen)
- Melissa Landrum, PhD (ClinVar)
- Brittany McLarney, MS, CGC (Invitae)
- Jud Rhode, BS (Invitae)
- Debbie Jae, MS, CGC (Invitae)
- ClinGen Steering Committee
- ClinGen Education WG
- The ClinVar Team

Learn more:

[http://bit.ly/GenomeConnect\\_](http://bit.ly/GenomeConnect_)



Questions?

[info@genomeconnect.org](mailto:info@genomeconnect.org)

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PMID: 30311371 and 26178529



**Thank you**

**Geisinger**

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# Supplemental Slides

\*\*\*,

Thank you for registering your \*\*\* with GenomeConnect in \*\*\*!

As part of your participation in GenomeConnect, you uploaded a copy of your genetic testing report so that we could collect information about any genetic changes that may have been found. You also indicated that you wished to receive updates regarding these results from the GenomeConnect team should we learn of any. **We are reaching out to let you know that there may be updated information about the genetic change in the \*\*\* gene listed on \*\*\* report dated \*\*\*. This updated result may or may not impact \*\*\* medical management.** You may have also already received this update if the \*\*\* report is not your most recent report.

To learn more about this update, we encourage you to reach out to the healthcare provider that ordered your testing or a genetics expert in your area. If you do not remember which provider ordered your genetic testing, our team may be able to help. Contact us by emailing [info@genomeconnect.org](mailto:info@genomeconnect.org) or by calling 570-214-1721. To find a genetic counselor in your area, you can use the following website: <https://www.nsgc.org/page/find-a-genetic-counselor>.

It may be helpful to share this email with the healthcare provider you meet with. It is also helpful for them to know \*\*\*'s testing was performed at \*\*\* and has the ID number \*\*\*. **Your healthcare provider can reach out to \*\*\* to discuss any updates to your genetic testing results and to request an updated report.** \*\*\* can be reached at LAB NUMBER. Your healthcare provider also can find helpful information on our [website](#) and can find more information about your variant on the ClinVar website (ClinVar Link).

If you receive an updated genetic test report, we ask that you upload this to your GenomeConnect account so that we can update our records.

Thank you again for your participation and commitment to advancing our understanding of genetics.

Best,

The GenomeConnect Team

[info@genomeconnect.org](mailto:info@genomeconnect.org)